GEPHE SUMMARY

Gephebase Gene
hemoglobin; HBB (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^hemoglobin; HBB^#gephebase-summary-title)

GepheID
GP00000468

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Physiology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Physiology^#gephebase-summary-title)

Trait
Hypoxia response (https://www.gephebase.org/search-criteria?/and+Trait=^Hypoxia response^#gephebase-summary-title)

Trait State in Taxon A
Anas georgica - low-altitude

Trait State in Taxon B
Anas georgica - high-altitude

Ancestral State
Taxon A

Taxonomic Status
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Intraspecific^#gephebase-summary-title)

Taxon A

Latin Name
Anas georgica (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Anas georgica^#gephebase-summary-title)

Common Name
yellow-billed pintail

Synonyms
yellow-billed pintail; Anas georgica Gmelin, 1789

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Archelosauria; Archosauria; Dinosauria; Saurischia; Theropoda; Coelurosauria; Aves; Neognathae; Galloanserae; Anseriformes; Anatidae; Anatinae; Anas

Parent
Anas (ducks) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8835)

NCBI Taxonomy ID
75847 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 75847)

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Anas georgica (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Anas georgica^#gephebase-summary-title)

Common Name
yellow-billed pintail

Synonyms
yellow-billed pintail; Anas georgica Gmelin, 1789

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Archelosauria; Archosauria; Dinosauria; Saurischia; Theropoda; Coelurosauria; Aves; Neognathae; Galloanserae; Anseriformes; Anatidae; Anatinae; Anas

Parent
Anas (ducks) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8835)

NCBI Taxonomy ID
75847 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 75847)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
HBB

Synonyms
ECYT6; CD113t-C; beta-globin

String
9606.ENSP00000333994 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000333994)

Sequence Similarities
Belongs to the globin family.

GO - Molecular Function
GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)
GO:0020037 : heme binding (https://www.ebi.ac.uk/QuickGO/term/GO:0020037)
GO:0005344 : oxygen carrier activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005344)
GO:0043177 : organic acid binding (https://www.ebi.ac.uk/QuickGO/term/GO:0043177)
GO:0019825 : oxygen binding (https://www.ebi.ac.uk/QuickGO/term/GO:0019825)
GO:0031721 : hemoglobin alpha binding

UniProtKB Homo sapiens
P68871 (http://www.uniprot.org/uniprot/P68871)

GenebankID or UniProtKB
ACM67760 (https://www.ncbi.nlm.nih.gov/nuccore/ACM67760)

(https://www.ebi.ac.uk/QuickGO/term/GO:0031721)
GO:0030492 : hemoglobin binding (https://www.ebi.ac.uk/QuickGO/term/GO:0030492)

GO - Biological Process

GO:0006898 : receptor-mediated endocytosis
(https://www.ebi.ac.uk/QuickGO/term/GO:0006898)
GO:0007596 : blood coagulation (https://www.ebi.ac.uk/QuickGO/term/GO:0007596)
GO:0008217 : regulation of blood pressure
(https://www.ebi.ac.uk/QuickGO/term/GO:0008217)
GO:0042542 : response to hydrogen peroxide
(https://www.ebi.ac.uk/QuickGO/term/GO:0042542)
GO:0043312 : neutrophil degranulation
(https://www.ebi.ac.uk/QuickGO/term/GO:0043312)
GO:0015701 : bicarbonate transport (https://www.ebi.ac.uk/QuickGO/term/GO:0015701)
GO:0098869 : cellular oxidant detoxification
(https://www.ebi.ac.uk/QuickGO/term/GO:0098869)
GO:0042744 : hydrogen peroxide catabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0042744)
GO:0015671 : oxygen transport (https://www.ebi.ac.uk/QuickGO/term/GO:0015671)
GO:0010942 : positive regulation of cell death
(https://www.ebi.ac.uk/QuickGO/term/GO:0010942)
GO:0051291 : protein heterooligomerization
(https://www.ebi.ac.uk/QuickGO/term/GO:0051291)
GO:0030185 : nitric oxide transport (https://www.ebi.ac.uk/QuickGO/term/GO:0030185)
GO:0070527 : platelet aggregation (https://www.ebi.ac.uk/QuickGO/term/GO:0070527)
GO:0045429 : positive regulation of nitric oxide biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0045429)
GO:0050880 : regulation of blood vessel size
(https://www.ebi.ac.uk/QuickGO/term/GO:0050880)
GO:0070293 : renal absorption (https://www.ebi.ac.uk/QuickGO/term/GO:0070293)

GO - Cellular Component

GO:0005829 : cytosol (https://www.ebi.ac.uk/QuickGO/term/GO:0005829)
GO:0070062 : extracellular exosome (https://www.ebi.ac.uk/QuickGO/term/GO:0070062)
GO:0005576 : extracellular region (https://www.ebi.ac.uk/QuickGO/term/GO:0005576)
GO:0005615 : extracellular space (https://www.ebi.ac.uk/QuickGO/term/GO:0005615)
GO:0072562 : blood microparticle (https://www.ebi.ac.uk/QuickGO/term/GO:0072562)
GO:0071682 : endocytic vesicle lumen
(https://www.ebi.ac.uk/QuickGO/term/GO:0071682)
GO:0031838 : haptoglobin-hemoglobin complex
(https://www.ebi.ac.uk/QuickGO/term/GO:0031838)
GO:0005833 : hemoglobin complex (https://www.ebi.ac.uk/QuickGO/term/GO:0005833)
GO:1904813 : ficolin-1-rich granule lumen
(https://www.ebi.ac.uk/QuickGO/term/GO:1904813)
GO:1904724 : tertiary granule lumen (https://www.ebi.ac.uk/QuickGO/term/GO:1904724)

Mutation #1

Presumptive Null

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Coding^#gephebase-summary-title)

Aberration Type

SNP (https://www.gephebase.org/search-criteria?/and+Aberration Type=^SNP^#gephebase-summary-title)

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Ala116Ser; Leu133Met

Experimental Evidence

Candidate Gene (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Candidate Gene^#gephebase-summary-title)

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Ala	Ser	116

Main Reference

Parallel evolution in the major haemoglobin genes of eight species of Andean waterfowl. (2009) (https://pubmed.ncbi.nlm.nih.gov/19754505)

Authors

McCracken KG; Barger CP; Bulgarella M; Johnson KP; Sonsthagen SA; Trucco J; Valqui TH; Wilson RE; Winker K; Sorenson MD

Abstract

Theory predicts that parallel evolution should be common when the number of beneficial mutations is limited by selective constraints on protein structure. However, confirmation is scarce in natural populations. Here we studied the major haemoglobin genes of eight Andean duck lineages and compared them to 115 other waterfowl species, including the bar-headed goose (Anser indicus) and Abyssinian blue-winged goose (Cyanochen cyanopterus), two additional species living at high altitude. One to five amino acid replacements were significantly overrepresented or derived in each highland population, and parallel substitutions were more common than in simulated sequences evolved under a neutral model. Two substitutions evolved in parallel in the alpha A subunit of two (Ala-alpha 8) and five (Thr-alpha 77) taxa, and five identical beta A subunit substitutions were observed in two (Ser-beta 4, Glu-beta 94, Met-beta 133) or three (Ser-beta 13, Ser-beta 116) taxa. Substitutions at adjacent sites within the same functional protein region were also observed. Five such replacements were in exterior, solvent-accessible positions on the A helix and AB corner of the alpha A subunit. Five others were in close proximity to inositolpentaphosphate binding sites, and two pairs of independent replacements occurred at two different alpha(1)beta(1) intersubunit contacts. More than half of the substitutions in highland lineages resulted in the acquisition of

serine or threonine (18 gains vs. 2 losses), both of which possess a hydroxyl group that can hydrogen bond to a variety of polar substrates. The patterns of parallel evolution observed in these waterfowl suggest that adaptation to high-altitude hypoxia has resulted from selection on unique but overlapping sets of one to five amino acid substitutions in each lineage.

Additional References

Convergent Evolution of Hemoglobin Function in High-Altitude Andean Waterfowl Involves Limited Parallelism at the Molecular Sequence Level. (2015) (https://pubmed.ncbi.nlm.nih.gov/26637114)

Mutation #2

Presumptive Null

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Coding^#gephebase-summary-title)

Aberration Type

SNP (https://www.gephebase.org/search-criteria?/and+Aberration Type=^SNP^#gephebase-summary-title)

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Ala116Ser; Leu133Met

Experimental Evidence

Candidate Gene (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Candidate Gene^#gephebase-summary-title)

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Leu	Met	133

Main Reference

Parallel evolution in the major haemoglobin genes of eight species of Andean waterfowl. (2009) (https://pubmed.ncbi.nlm.nih.gov/19754505)

Authors

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Abstract

Theory predicts that parallel evolution should be common when the number of beneficial mutations is limited by selective constraints on protein structure. However, confirmation is scarce in natural populations. Here we studied the major haemoglobin genes of eight Andean duck lineages and compared them to 115 other waterfowl species, including the bar-headed goose (Anser indicus) and Abyssinian blue-winged goose (Cyanochen cyanopterus), two additional species living at high altitude. One to five amino acid replacements were significantly overrepresented or derived in each highland population, and parallel substitutions were more common than in simulated sequences evolved under a neutral model. Two substitutions evolved in parallel in the alpha A subunit of two (Ala-alpha 8) and five (Thr-alpha 77) taxa, and five identical beta A subunit substitutions were observed in two (Ser-beta 4, Glu-beta 94, Met-beta 133) or three (Ser-beta 13, Ser-beta 116) taxa. Substitutions at adjacent sites within the same functional protein region were also observed. Five such replacements were in exterior, solvent-accessible positions on the A helix and AB corner of the alpha A subunit. Five others were in close proximity to inositolpentaphosphate binding sites, and two pairs of independent replacements occurred at two different alpha(1)beta(1) intersubunit contacts. More than half of the substitutions in highland lineages resulted in the acquisition of serine or threonine (18 gains vs. 2 losses), both of which possess a hydroxyl group that can hydrogen bond to a variety of polar substrates. The patterns of parallel evolution observed in these waterfowl suggest that adaptation to high-altitude hypoxia has resulted from selection on unique but overlapping sets of one to five amino acid substitutions in each lineage.

Additional References

Convergent Evolution of Hemoglobin Function in High-Altitude Andean Waterfowl Involves Limited Parallelism at the Molecular Sequence Level. (2015) (https://pubmed.ncbi.nlm.nih.gov/26637114)

RELATED GEPHE

Related Genes

2 (EGLN1, EPAS1) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^75847^/and+Trait=Hypoxia response/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

Needs curation @SeveralMutationsWithEffect